MEDGENOME

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GeKNOWme

Internal Quarterly Newsletter

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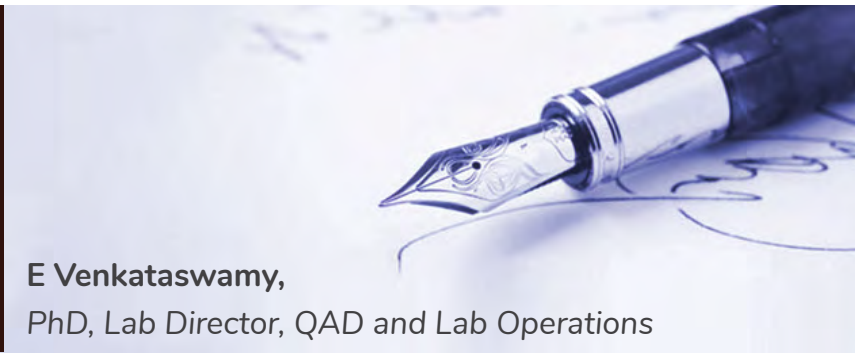


E Venkataswamy,
PhD, Lab Director, QAD and Lab Operations

Dear MedGenome Family,

Greetings!

I have completed almost 8 years in MedGenome and what a journey it has been so far! I have been extended wonderful opportunities at MedGenome and the learning has never stopped. At this juncture, I would like to take a moment to thank all of you for making the lab feel just like home.

I admire Dr. Ram and Dr. Sakthi's leadership skills and I am grateful to them for instilling faith in me and supporting me in my career growth at MedGenome. I started my journey at MedGenome as a Senior Scientist and then advanced to the role of the Associate Director of QA and subsequently to the role of the Lab Director. This has been an exciting journey for me as I have gained enormous knowledge in the various fields of NGS, Prenatal, Sanger, Microarray and Quality Assurance.

"Your work is going to fill a large part of your life, and the only way to be truly satisfied is to do what you believe is great work. And the only way to do great work is to love what you do."

- Steve Jobs

I always scored a centum in Math during my school days and my peers thought I would explore the path of Mathematics and contribute enormously to the field. But I chose a different path, and I love what I do, and it has taken me into different avenues and has helped me achieve great heights.

MedGenome has created a mark in the Genomics space owing to its clear vision fuelled by inspiring leadership and hard work of its leaders. It is an honour to work with the MedGenome team. Our goal for the coming years will be to make MedGenome as one of the most favoured labs by our customers. To achieve this goal, we need to focus on providing exceptional customer service and to adhere to the stringent quality norms along with cost-effective laboratory services.

As one of the leading companies in Genomics and Clinical Diagnostics, we want to provide our clients with the best quality and service. For this purpose, we volunteered to have third-party audits to assess our quality process and systems. We worked towards CAP accreditation, and we have had this accreditation for the last 4 years and we will continue to maintain it in future too. Recently, we got 2 more accreditations- NABL accreditation for COVID-19 testing and an accreditation from EFI for histocompatibility testing. Our company spends huge effort in proficiency testing to ensure we provide best results to the patients and clients. We are following good lab practices and now we are focusing on good documentation practices and good clinical practices so that we can strengthen our offerings in clinical research areas.

We have bi-weekly meetings with all department leads to discuss the issues affecting quality. The leads present the performance and the trend of quality indicators (TAT, repetitions, revision of reports, etc.)

specific to their domains. We have monthly PT evaluation meetings where we discuss the results received from CAP/ILC/EQAS which help in improving the lab protocols, workflows, and analytical pipelines.

As part of continuing education, we encourage everyone to attend internal journal clubs to keep abreast with the knowledge. Every week we have stimulating discussions on various intriguing topics related to the techniques used in our lab. We encourage our team members to participate in various webinars related to our field to expand their knowledge.

To ensure a continuous improvement in lab quality, our focus will be towards judicious expenditure on all lab consumables, maintaining inexhaustive but valid and right stock, release reports within defined TAT, zero amended reports, addition of new tests, scoring 100% in proficiency testing, maintenance of accreditation with reduced or no nonconformities, reduction of repeats and providing seamless tech support.

These unprecedented times have been both a challenging and a learning experience for all of us. With our ultimate dedication, strong team support, high quality infrastructure and well-established processes, I am proud that we were able to make a difference in people's lives.

> **"All talk is a lie in a way. Only the doing of a thing can make it true"**
>
> – Donal Ryan

Contents

Most Talked About

The News

Diagnosing Lung Cancer at Ar Can Save Lives, Know About Testing Lung Cancer

Lung cancer is one of the common cancers in India. Early diagnosis can prevent lung ca

Updated at: Aug 08, 2021 Verified by DOCTOR

Listen to this article now 04:42 Powered by Trinity Audio

CANCER
Written by: Chanchal Sengar
Published at: Aug 08, 2021

In the past few decades, we all have seen multiple advancements in healthcare and many novel treatments have been discovered to manage cancer, however Lung cancer is still the leading cause of cancer related deaths in the entire world. It accounts for more death each year than breast, colon and prostate cancers combined. On an average it accounts for approximately 1.80 million deaths annually, according to the World Health Organisation (WHO). In this article, **Dr. G Arun Kumar, Head of Technical Support and Scientific Affairs Manager in Oncology, MedGenome labs** explains different methods of diagnosing lung cancer at the initial level.

Tobacco- The main cause of lung cancer in India

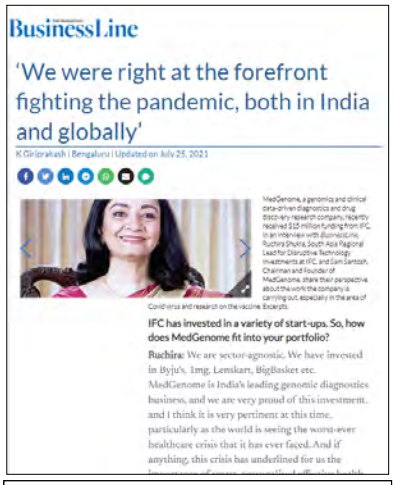
Forbes LEADERSHIP INNOVATION BILLIONAIRES LISTS STARTUPS PODCASTS

Inside the world of DNA decoding in India

Even as genetic-sequencing technology becomes cheaper and faster, the adoption of precision medicine, which decodes a patient's DNA to treat them in a more personalised, targeted way, remains limited in India. What will it take to turn it into an everyday reality?

Scientists working on genome sequencing at the MedGenome lab in Bengaluru. Image: Himadri Sharma for Forbes India

By the time Devina Bose, 65, noticed a lump in her breast, the tumour had advanced to stage four. Tests revealed that the cancerous cells were hormone receptor positive, which means they needed estrogen and progesterone to grow and spread, but HER2 negative, referring to a growth-promoting protein found on



BusinessLine

'We were right at the forefront fighting the pandemic, both in India and globally'

K Giriprakash | Bengaluru | Updated on July 25, 2021

MedGenome, a genomics and clinical data-driven diagnostics and drug discovery research company, recently received $15 million funding from IFC. In an interview with *BusinessLine*, Ruchira Dhaka, South Asia Regional Lead for Disruptive Technology Investments at IFC, and Sam Santosh, Chairman and Founder of MedGenome, share their perspective about the work the company is carrying out, especially in the area of Covid virus and research on the vaccine. Excerpts:

IFC has invested in a variety of start-ups. So, how does MedGenome fit into your portfolio?

Ruchira: We are sector-agnostic. We have invested in Byju's, Img, Lenskart, BigBasket etc. MedGenome is India's leading genomic diagnostics business, and we are very proud of this investment, and I think it is very pertinent at this time, particularly as the world is seeing the worst-ever healthcare crisis that it has ever faced. And if anything, this crisis has underlined for us the

Bengaluru firm takes genome sequencing to people's homes

Bengaluru: MedGenome Labs, a Bengaluru-based genomics-driven research and diagnostics company, has launched a direct-to-consumer service, offering cutting edge technology like next-generation sequencing at the doorsteps of consumers.

Genessense — the new category introduced by MedGenome — focuses on rs-modelling the genomics space with evidence-backed tests that provide insights into a person's health much before the onset of symptoms.

VL Ramprasad, CEO, MedGenome, told **TOI**: "We had been thinking about this for about a year now but the pandemic caused some delay. We've launched two types of tests: One for CAD (coronary artery disease) and another for drug metabolising and risk profiling. People can start ordering tests for a cost."

He said that within the next year, the firm would add at least four more tests that could deal with diabetes, hypertension and more.

Ramprasad said the KARDIOGEN risk score (KRS), above 18 years of age. It not only predicts the inherited risk of heart disease, but also gives an opportunity to manage the disease. "It predicts the early onset of CAD with almost 90% accuracy," Ramprasad said.

The test helps predict one's chances of developing a heart condition even before the body shows any symptoms and puts a number to this risk — the KARDIOGEN risk score (KRS).

"KRS is determined through an assessment of variations in more than six million genes," the firm said.

"The association of all these unique variations gives us a polygenic risk score which we call the KRS. Based on KRS, one is classified as 'high, moderate or average risk', an insight that will help one take preventive action," the firm said.

— is a precision pharmacogenomics test for individuals that checks for metabolism of more than 45 drugs and drug classes. CUREGEN's results will let one know if they respond differently to these drugs and if they need to be aware that some of these drugs may cause an adverse reaction based on their genomic profile. This is also a test that a person needs to take only once in their life.

While insisting Genessense follows the latest guidelines prescribed by the Clinical Pharmacogenetics Implementation Consortium (CPIC) and PharmaGKB covering more than 45 drugs/drug classes across 10 disease categories, the firm said CUREGEN only looks at drugs for which level of evidence is strong as per established guidelines.

"CUREGEN leverages microarray/sequencing and CUREGEN Advanced leverages next-generation sequencing (NGS) to arrive at results. NGS picks up more than 250 critical variants that are missed by the older technique. MedGenome – Genessense's parent firm — was the first to

Meds for Each Other, P

Sam Santhosh & Shalmali Sardesai

How often do we look for medicines that are commonly used to treat a particular disease? What if we looked for medicines to treat a particular individual instead? Transitioning from the current one-cure-fits-all treatment model to a holistic approach that tailors the medical interventions to a particular individual is what precision medicine is all about.

At the launch of the US Precision Medicine Initiative in 2015, Barack Obama had said, "We can match a blood transfusion to a blood type — that was an important discovery. What if matching a cancer cure to our genetic code was just as easy, just as standard?" Personalised treatments benefit health by avoiding wrong treatment and reducing the financial burden for those who can't afford it. With an almost 72 million rare disease population in India, the right therapy — or preventing the occurrence itself — will go a long way to reduce the country's health burden.

The Human Genome Project started in 1990. Sequencing the human genome for the first time took 13 years, costing about $3 billion. Since then, sequencing technologies have undergone revolutionary changes, from sequencing one gene in a panel of genes by next-generation sequencing (NGS). The speed and cost efficiency of sequencing a whole human genome, a major driver of 'precision medicine, has improved manifold — from $3,000 in 2013 to about $1,000 today.

Understanding the genetic footprints of the patients is where genomic-based diagnostics comes in. Imagine the relief when an expecting couple with a firm born confirms from hearing and speech loss, gets to know through genetic testing of the fetus that their next born child would be a carrier of the disease, but wouldn't suffer from hearing loss. While prenatal testing detects genetic defects in a foetus before birth, there are various other clinical applications within reproductive genetics, such as carrier testing, pre-implantation testing and new-born screening.

Cancer, largely a genetic disease, has been the first and most widely studied disease area in precision

PMS will get personal

medicine, where diagnostics serves not as a one-time visit, but rather as ongoing process to assess the biology of the tumour as it changes and evolves. About 5-10% of all cancers are hereditary and occur due to inheritance of a genetic variation, mutation within families.

In 2021, World Health Organisation (WHO) projected that 1 in 10 Indians will develop cancer during their lifetime, and 1 in 15 will die of the disease. Identifying genetic predispositions can have significant implications for treatment decisions, interventions for patients and testing for close relatives.

Personalised cancer medicine also involves targeted therapy, which targets specific genes and proteins that allow certain cancers to grow and survive. Majority of cancers are also being successfully treated by immunotherapy, which boosts the body's natural defences to fight cancer. Though it remains very costly.

Another breakthrough — liquid biopsy — has opened doors to a new era in precision oncology. A simple blood test can detect cancer much earlier, reduce the burden on the limited cancer hospitals, while saving many more lives at much lower costs.

While we can't change our genes (yet), we can change the environment we put them in, and make healthier lifestyle choices. Medicine-based genomic tests combined with the analysis of other risk factors can help tailor specific nutritional and lifestyle choices. With pharmacogenomics, it is now possible to identify

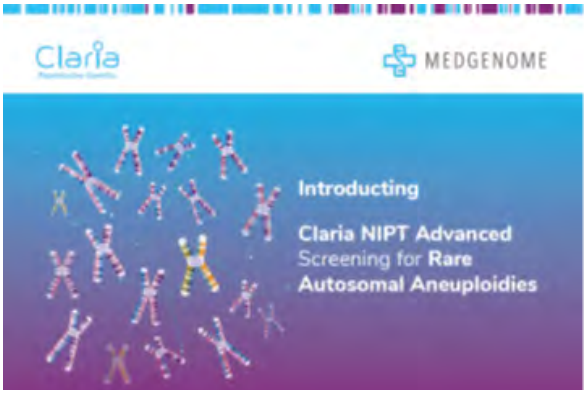
For press articles, please click https://diagnostics.medgenome.com/press/

MedGenome Connect

Claria
Reproductive Genetics

The quarter after the lockdown saw the continuing trend of increased adoption of NIPT. We further augmented our NIPT offering by launching our Rare Autosomal Aneuploidy (RAA) offering called Claria NIPT Advanced. RAA's are frequently associated with adverse pregnancy outcomes and are identified by Genome wide screening of cell free DNA. This was well received in the market and we are the only company that has validated RAA screening in India, another feather in our cap.

We continue to do more webinars during this quarter but can notice the increasing trend of webinar fatigue creep in. But as the lockdowns have eased and people have started coming out, the season for in person CME's and conference have started. We are looking at smaller towns and cities for our CME's. Our Dr. Priyadarshini Pandey and the local sales team received a very warm reception when they went to Latur in Maharashtra for a CME with the Latur ObGyn society. More such CME's are planned and we will also be engaging FOGSI and the local associations for the joint webinars in the coming quarter.



ACTIA
Inherited Genetics

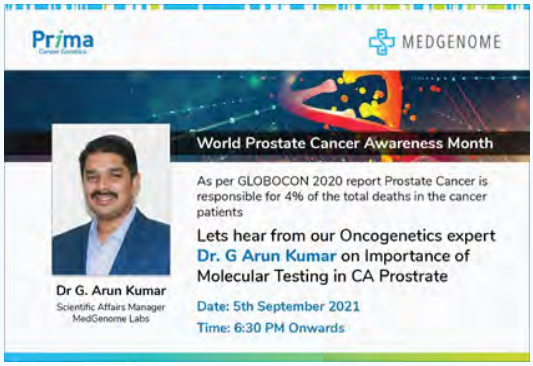
The post lockdown quarter was much better for Actia with OPD's coming back. The samples increased and so did the engagements. Two new publications were published in this quarter. The first one in association with NIMHANS looked at an extremely rare condition called Nemaline Rod/Cap Myopathy. The 2nd was about the experience of holding cardiogenetic clinics in tertiary care settings.

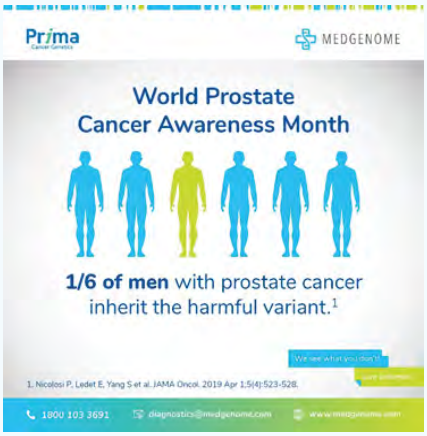
We also tied up with a crowd funding agency Impact Guru to support our Rare Disease patients with the option of raising funds through crowd funding. This initiative is part of our CSR efforts.

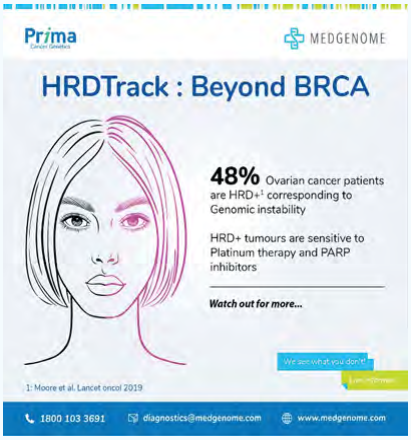
MedGenome Connect

Prima
Cancer Genetics

We continued to focus on our digital media for Prima. World Lung cancer day was celebrated on August 1, for which we did a digital campaign. Since September was a very important month from an oncology perspective as it is both Prostate Cancer awareness and Leukemia awareness month, we did a series of campaigns. The team is also gearing up to launch HRDTrack and a campaign has also been initiated for the same

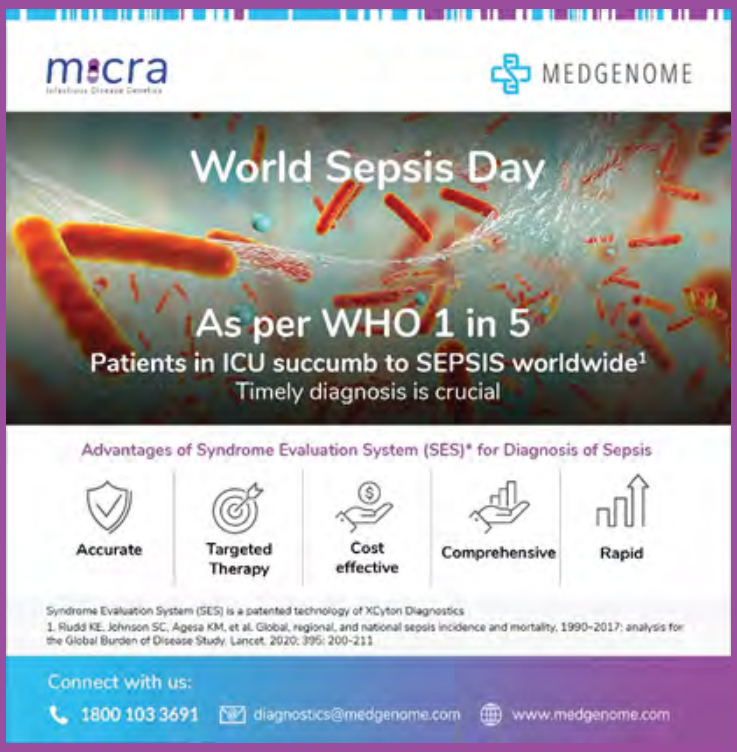






micra
Infectious Disease Genetics

The third quarter has been a very busy one for Micra. We ramped up our COVID RT-PCR testing as there were speculations about the new C1.2. strain of SARS COV2. Dr. Ram had an interaction with NDTV on the same. For the World Sepsis Day on September 14, a digital campaign was executed.



What's new

Publications

1. *Nemaline Rod/Cap Myopathy Due to Novel Homozygous MYPN Mutations: The First Report from South Asia and Comprehensive Literature Review- Published in Journal of Clinical Neurology*

   To read, click- https://thejcn.com/DOIx.php?id=10.3988/jcn.2021.17.3.409

2. *Initial experience and results of a cardiogenetic clinic in a tertiary cardiac care center in India- Published in Annals of Pediatric Cardiology*

   To read, click- https://www.annalspc.com/text.asp?2021/14/3/443/324640

3. *Immunodominant T-cell epitopes from the SARS-CoV-2 spike antigen reveal robust pre-existing T-cell immunity in unexposed individuals- Published in Nature*

   To read, click- https://www.nature.com/articles/s41598-021-92521-4

Proud moment

MedGenome has been awarded the Best Standalone Diagnostics Company (South) by ***ET Healthcare Awards 2021***

From our US office

As an ongoing effort to disseminate our scientific expertise in novel sequencing applications we recently added a new poster titled “MedGenome Inc. Broadens Single Cell Transcriptome and Epigenome Profiling: From Tissues to single nuclei RNA (snRNA) Sequencing and Data Analysis”. Please click here to know more:

https://research.medgenome.com/posters/

MedGenome Inc. continues to collaborate with customers on publications of impact, the latest one was published with Beaumont University on "Maternal opioid use disorder: Placental transcriptome analysis for neonatal opioid withdrawal syndrome" in "Genomics, Volume 113, Issue 6, 2021, Pages 3610-3617, ISSN 0888-7543"

To know more click on the link: ***https://research.medgenome.com/publications/***

We continue to add more articles on our Research Blog. MedGenome colleagues are encouraged to take initiative and contribute towards the blog. You can share your viewpoints and articles with Vinay and Hiran at:

mgus-blog@medgenome.com

To read our interesting articles on the cutting-edge research please visit us at:

https://research.medgenome.com/blog/

The recent ones added talks about: “High-throughput antibody discovery using single-cell B-cell receptor sequencing (scBCR-seq) “HitMab””

Also, MedGenome US conducts regular internal trainings for Commercial and NGS lab members on assays offered. One of them on 'Single cell Sequencing' is available in the link below.

https://youtu.be/GktwPX4OIbw

The recordings are archived in LIMS for all the team members to review at any time.

Thalassemia – a story of love, hope and perseverance

Ramdev hailed from Orai, a small agricultural village in the district of Jalaun, Uttar Pradesh. He owned a piece of land and the income from his farming activities was enough to run his family that consisted of his wife Savita, two-year old son Sumit and six months old daughter Tanya. Life was content until the young couple started to observe significant changes in their son's growth. The child displayed weakness and fatigue, exhibited slow growth and abdominal swelling and pale skin. Although the young parents tried to convince themselves that nothing was wrong with their son, parental instincts kicked in and they took their child to a primary health centre in the nearby town from where they were directed to Government medical college hospital in Jhansi.

That is where the couple learnt that their son was afflicted with a blood disorder called Thalassemia, that it was a hereditary disorder and that as part of treatment, he would require periodic blood transfusions. From what little they understood about the disease they hoped against hope that their young daughter would be spared. However, by the time Sumit needed regular transfusions, Tanya also was diagnosed with the same condition and thus a nightmare began for the parents of having to care for two ailing children.

The couple approached an NGO referred by their doctor in Jhansi and with their help registered their children with a patient welfare society attached to a hospital that provided treatment for thalassemia patients at a nominal cost. Sumit and Tanya fought their condition bravely through their growing up years, pursued their education and made a mark for themselves.

Thalassemia is an autosomal recessive disorder and is characterized by defective formation of hemoglobin, the component present in red blood cells that is responsible for the transport of oxygen to the cells. The hemoglobin is made of four globin molecules – two alpha chains and two beta chains and a heme molecule. Two genes, namely HBA1 and HBA2 make alpha globin chains and HBB gene is responsible for the formation of beta chains. All genes are present in two copies, one inherited from the father and the other from the mother. Variations in the sequence, known as mutations, in both copies of either HBA1 and HBA2 genes result in alpha thalassemia. Similarly, mutations in HBB gene result in beta thalassemia. Autosomal recessive disorders, like Thalassemia, occur either because both parents carry one copy each of the mutated gene and pass it on to their offspring or because of new mutation in an individual.

Sumit and Tanya came to know that both their parents were carriers of a defect in HBB gene and they unfortunately inherited the defective genes from both their parents. Over the years much has been discovered about the heritability of the disease and now tests are available to identify if parents carry a defective gene that can result in the disease condition in their baby. This is called carrier testing.

Carrier testing can help in identifying if both parents of a child with an autosomal recessive disorder carry the defective gene. It also helps in counselling of couples looking to marry in consanguinity or have a history of recessive disorders in their families. MedGenome Labs Ltd offers carrier testing for various recessive disorders and also provides genetic counselling services. A comprehensive genetic counselling that includes a detailed discussion with a healthcare provider to understand the nature of genetic disease, coupled with relevant genetic testing can go a long way in preventing inherited disorders.

Antimicrobial resistance:

A silent pandemic

Gunisha Pasricha, PhD
Principal Scientist

Introduction

Antimicrobials include antibiotic, antiviral, antifungal and antiparasitic medicines which fight against bacteria, viruses, fungi, and parasites respectively. WHO has declared that antimicrobial resistance (AMR) is one of the top 10 global public health threats facing humanity which require immediate multi-sectoral actions. AMR occurs when microbes resist the effects of medicines and develop the ability to defeat the drugs designed to kill them- hence they are called 'super-bugs'. This makes common infections harder to treat and increases the risk of disease spread, severe illness and death. According to a recent estimate, ~700,000 people die every year globally from drug-resistance infections, and the number is projected to increase to 10 million by 2050.[1] This would mean that AMR would surpass cancer as a major cause of death worldwide.[2]

Major drivers of AMR

Major driver of AMR is the overuse and misuse of antimicrobials in humans and animals. Inappropriate prescribing of anti-microbials, lack of sanitation and hygiene, poor infection and disease prevention, suboptimal rapid diagnostics and vaccines, extensive use of antibiotics in agriculture also contribute significantly to spread of AMR (Figure 1).[3]

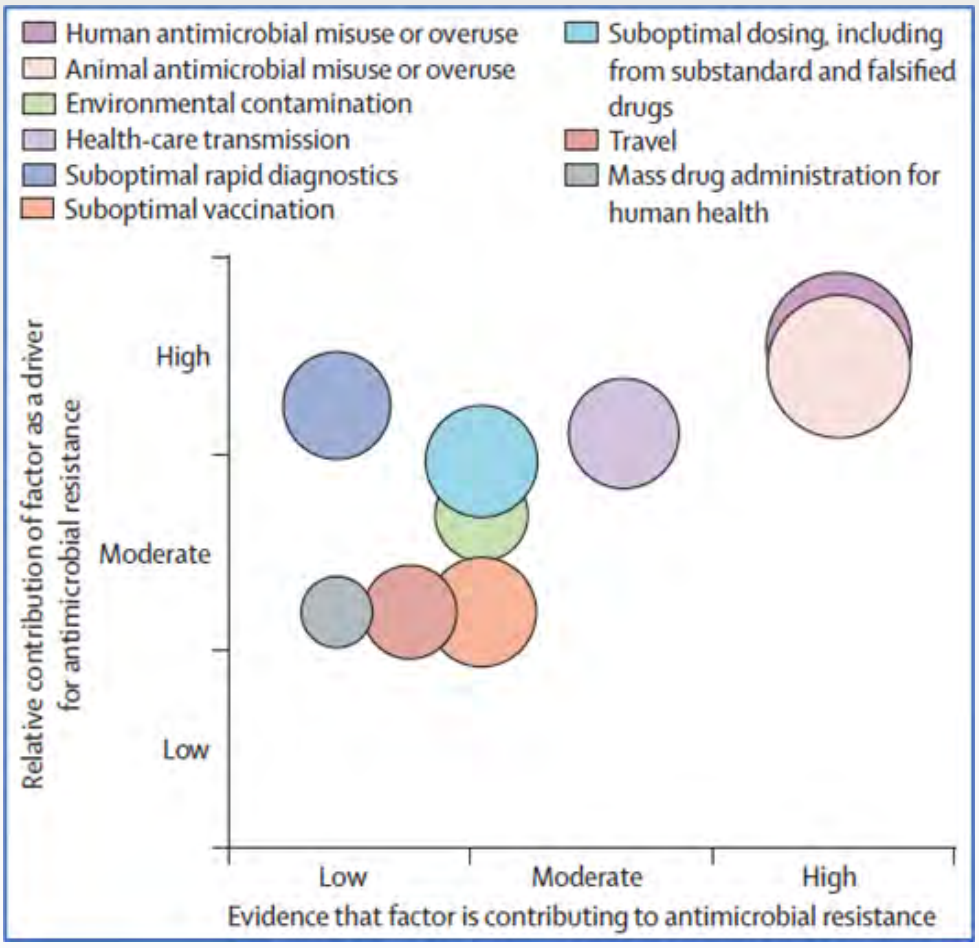


Figure 1: Drivers of antimicrobial resistance[3]

Bacterial AMR

Although drug resistance in bacteria is a global threat, but nowhere is it as stark as in India.[4] The WHO India Country Cooperation Strategy 2019-2023 recognizes containment of antimicrobial resistance as a priority.[5] Indian Priority Pathogen List (IPPL) in 2019 grouped the bacterial pathogens associated with antibiotic resistance according to the species and resistance and further stratified into three priority tiers – critical, high and medium (Table 1). Mycobacteria *spps*. (including Mycobacterium tuberculosis) were not included in this prioritization exercise as it is a well-established global and national priority for which innovative new treatments are urgently needed and being developed.[5]

Table 1: Indian priority pathogen list[5]

CRITICAL PRIORITY	**Enterobacteriaceae** *(Klebsiella pneumoniae and Escherichia coli)*	Carbapenem – R Tigecycline – R Colistin – R
	Non-fermenting bacteria *(Acinetobacter baumannii and Pseudomonas aeruginosa)*	Carbapenem – R Colistin – R
HIGH PRIORITY	**Staphylococcus aureus**	MRSA, hVISA Daptomycin – NS Linezolid – R
	Enterococcus species	Vancomycin – R Linezolid – R Daptomycin – NS
	Salmonella species *(Typhoidal and Non-typhoidal)*	Azithromycin – NS Third generation cephalosporins – NS Carbapenem – NS
MEDIUM PRIORITY	**Streptococcus pneumoniae**	Cephalosporin – R Fluoroquinolones – R Linezolid – R
	Staphylococcus, coagulase-negative	Vancomycin – R Linezolid – R
	Shigella species	Third generation cephalosporins – R Azithromycin – R
	Haemophilus influenzae	Third generation cephalosporin – NS Carbapenem – NS
	Neisseria meningitidis	Fluoroquinolones – NS Third generation cephalosporins – NS

Biology of bacterial AMR

Bacterial AMR has both genetic (stable and heritable) and phenotypic forms (reversible and not attributable to a genetic change). The clinical challenge of genetic AMR is enormous. The clinical recognition and experimental analysis of genetic AMR are relatively straightforward. In contrast, phenotypic AMR is much harder to recognize clinically and analyze experimentally. Phenotypic AMR can be expressed by a subset of bacterial cells that have a much slower rate of killing than most of the population or are not killed at all. This is often called "persistence" or also sometimes termed "tolerance"(Figure 2).[6]

In genetic AMR, exposure to antibiotics eliminates drug-susceptible competitors, allowing the resistant bacteria (resister) to survive and multiply. Because bacteria grow and multiply fast, the resistant variants quickly dominate the population (Figure 2).

Phenotypic AMR can arise stochastically, or it can arise upon exposure to altered environments, such as nutrient or oxygen deprivation, acidification, oxidative stress, host immune responses, and exposure to sublethal concentrations of antibiotics. The distinction between genetic and phenotypic resistance is not always clear. Our scientific understanding of genetic AMR is robust; however, it is still nascent for phenotypic resistance.[6]

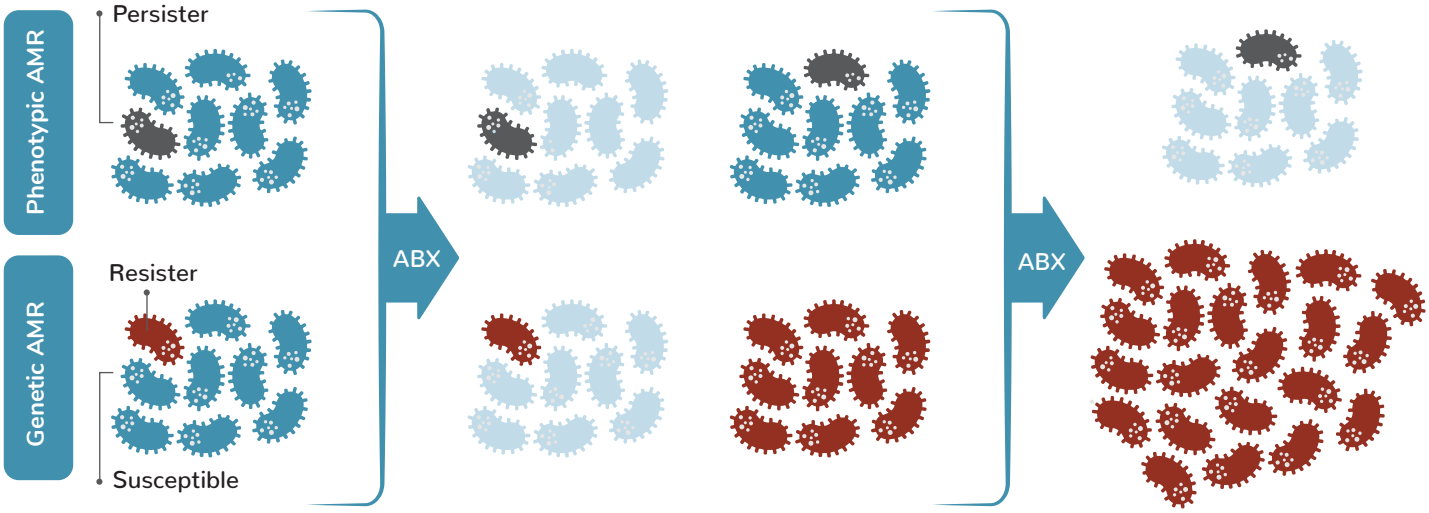


Figure 2: Genetic and phenotypic AMR[6]

Genetic bacterial AMR

Bacteria have the natural ability to evolve and adapt to their environment by changing their genetic material as a survival mechanism. Antibiotics disrupt essential cellular structures or biosynthetic pathways to kill bacteria or stop them from multiplying. As a response, bacteria have developed the following strategies to develop drug resistance, i.e., remove the antibiotics using an efflux pump; destroy the antibiotics by producing an inactivating enzyme; produce an alternative protein to bypass inhibition; modify the cellular component to circumvent being a target; restrict antibiotics access by changing the entry ways (Figure 3a). All of the survival mechanism are genetically encoded which can occur by overexpression or duplication of existing genes, point mutations or the acquisition of entirely new genes via horizontal gene transfer (HGT)(Figure 3b).[7]

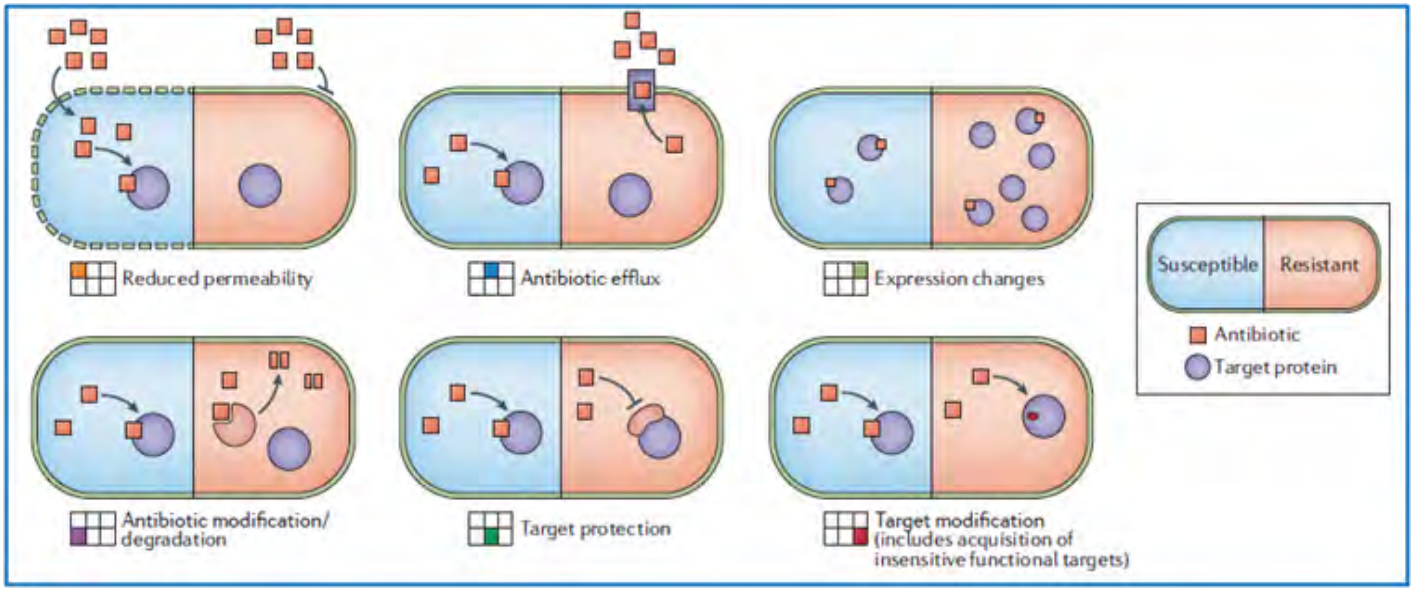


Figure 3a: Mechanism of antibiotic resistance and 3b: Genetic determinations of drug resitance[7]

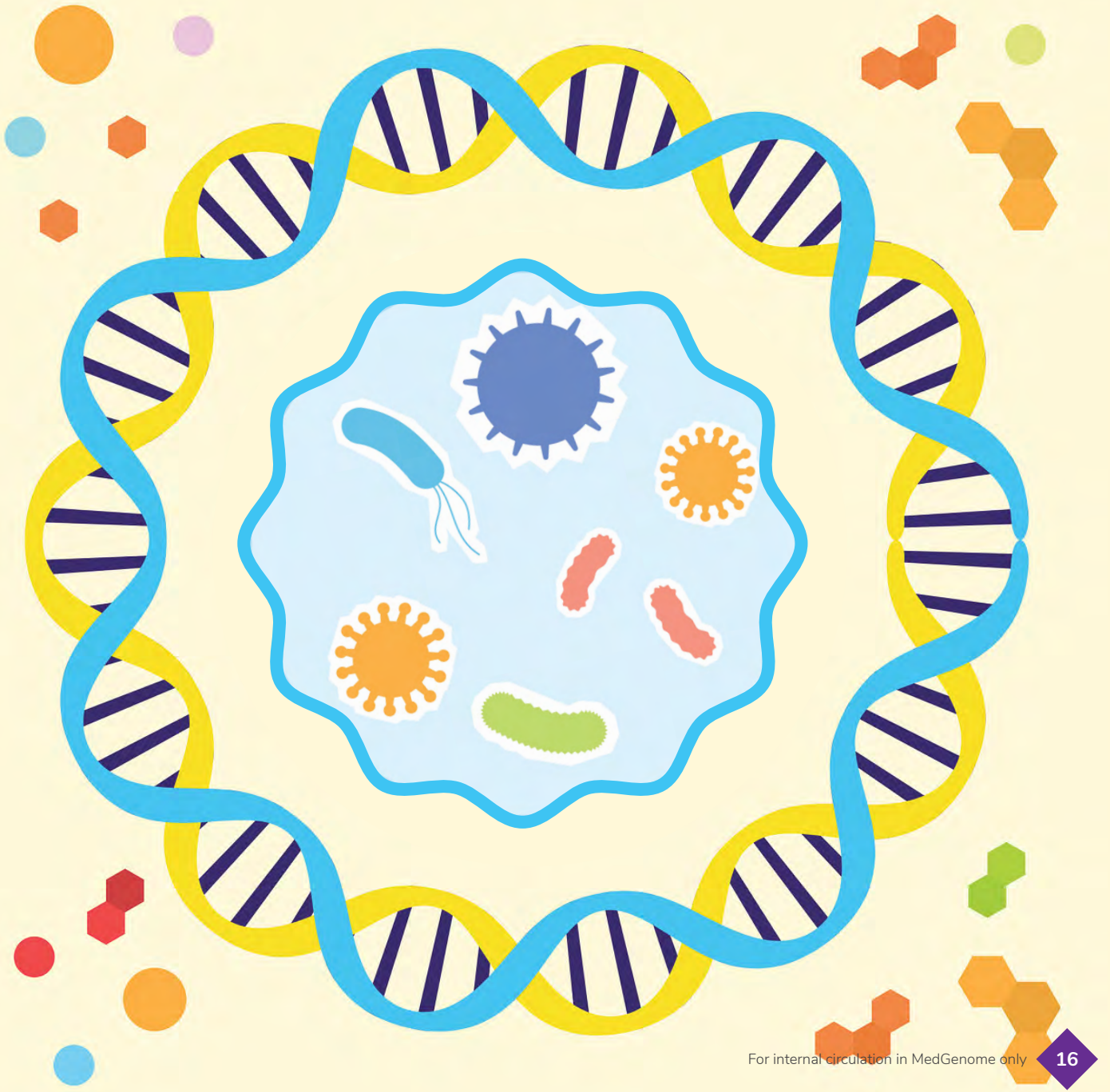
Bacterial AMR and infections

For common bacterial infections, including urinary tract infections, sepsis, sexually transmitted infections, and some forms of diarrhea, high rates of antibiotic resistance have been observed world-wide, indicating that we are running out of effective antibiotics. Antibiotic resistant Mycobacterium tuberculosis strains are threatening progress in controlling the global tuberculosis epidemic. Multidrug resistant tuberculosis (MDR-TB) requires treatment courses that are longer, less effective and far more expensive than those for non-resistant TB. Less than 60% of those treated for MDR/RR-TB are successfully cured.[1] The battle between antimicrobial-resistant pathogens and antibiotic therapy is an evolutionary arms race —one that we are currently losing. According to WHO, the clinical pipeline of new antimicrobials is dry.[1]

Next generation sequencing and AMR

In the fight against AMR, genome-based techniques can play a pivotal role. These tests have the promise to overcome the shortfalls of conventional techniques like culture- which is time consuming and laborious. Culture-based antimicrobial susceptibility testing is still the primary method employed by clinical laboratories to test antimicrobial resistance. Technologies such as next-generation sequencing (NGS) are expanding our abilities to detect and study antimicrobial resistance. Methods for predicting AMR genetic determinants from NGS data rely on complex bioinformatics algorithms and procedures to transform the large output produced by the sequencing technologies into relevant information. Whole-genome sequencing for antimicrobial susceptibility testing offers the potential to provide rapid, consistent, and accurate predictions of every known resistance phenotype for a strain, as well as simultaneously provide rich surveillance data.[7]

In tuberculosis, the biggest challenge is multi-drug resistance, hence last year, we launched our proprietary SPIT-SEQ test, a direct sputum based whole genome sequencing test for diagnosis and drug resistance profiling in pulmonary tuberculosis patients. We are also validating a NGS based Pan-Pathogen test with a select AMR panel to detect anti-bacterial, anti-viral and anti-fungal drug resistance directly in clinical samples. HIV and CMV genetic drug resistance NGS based tests are also being validated and would be soon launched.



Conclusions

Increased vaccination coverage, infection control and prevention, optimal diagnostic techniques and clean water and sanitation can reduce the need for antibiotics and in turn reduce spread of AMR. Also, stewardship programs at global are required to promote the judicious use of antimicrobials across all sectors. Research on both genetic and phenotypic AMR is warranted and specifically so in phenotypic AMR since it is more widespread and unrecognized till recently due to lack of appropriate techniques. More recently, NGS based whole genome and metagenome sequencing methods have been employed to detect genetic AMR determinants in both clinical and environmental samples and we believe that the story about NGS and AMR is just at its beginning.

AMR is a silent pandemic- it spreads globally through international travel and migration; however, its impact is felt slowly with long-term implications for global health. At present the world is dealing with acute ravages of COVID-19 and the collateral damage on other public health threats like AMR is apparent and imminent. A surge in use of antibiotic, antiviral, anti-parasite, and anti-inflammatory drugs during the COVID-19 pandemic has disrupted stewardship practices which were initiated in past decade to curb and control AMR spread thus accelerating the evolution and spread of AMR.[10]

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Matrilineal analysis of mutations in the DMD gene in a multigenerational South Indian cohort

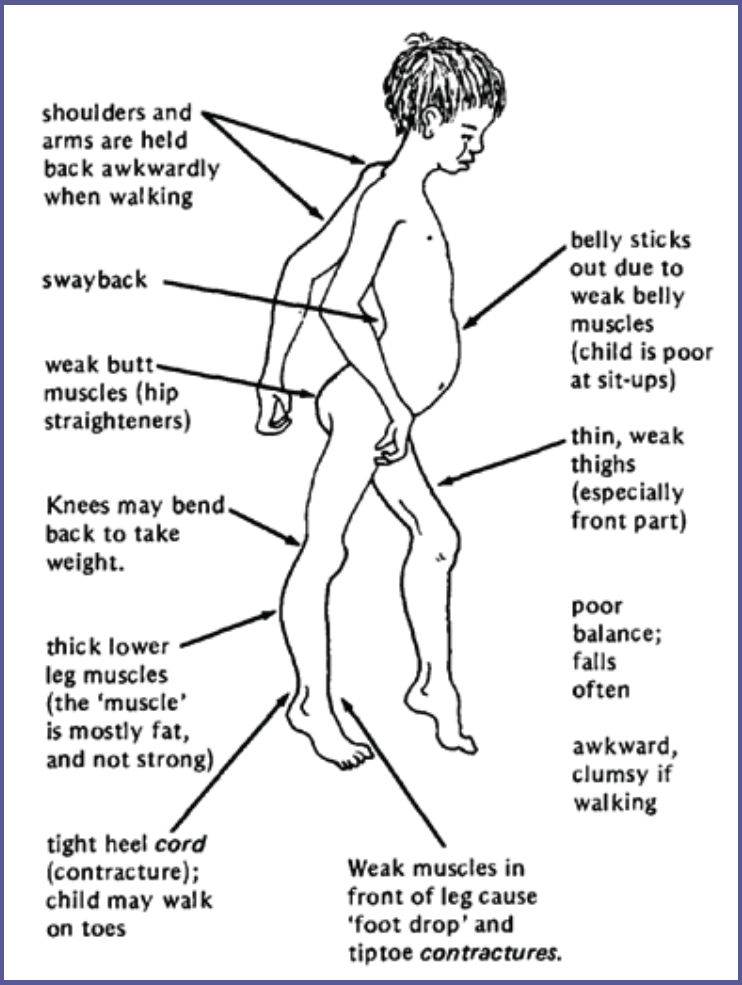
Ravi Gupta, PhD,
Chief Scientist

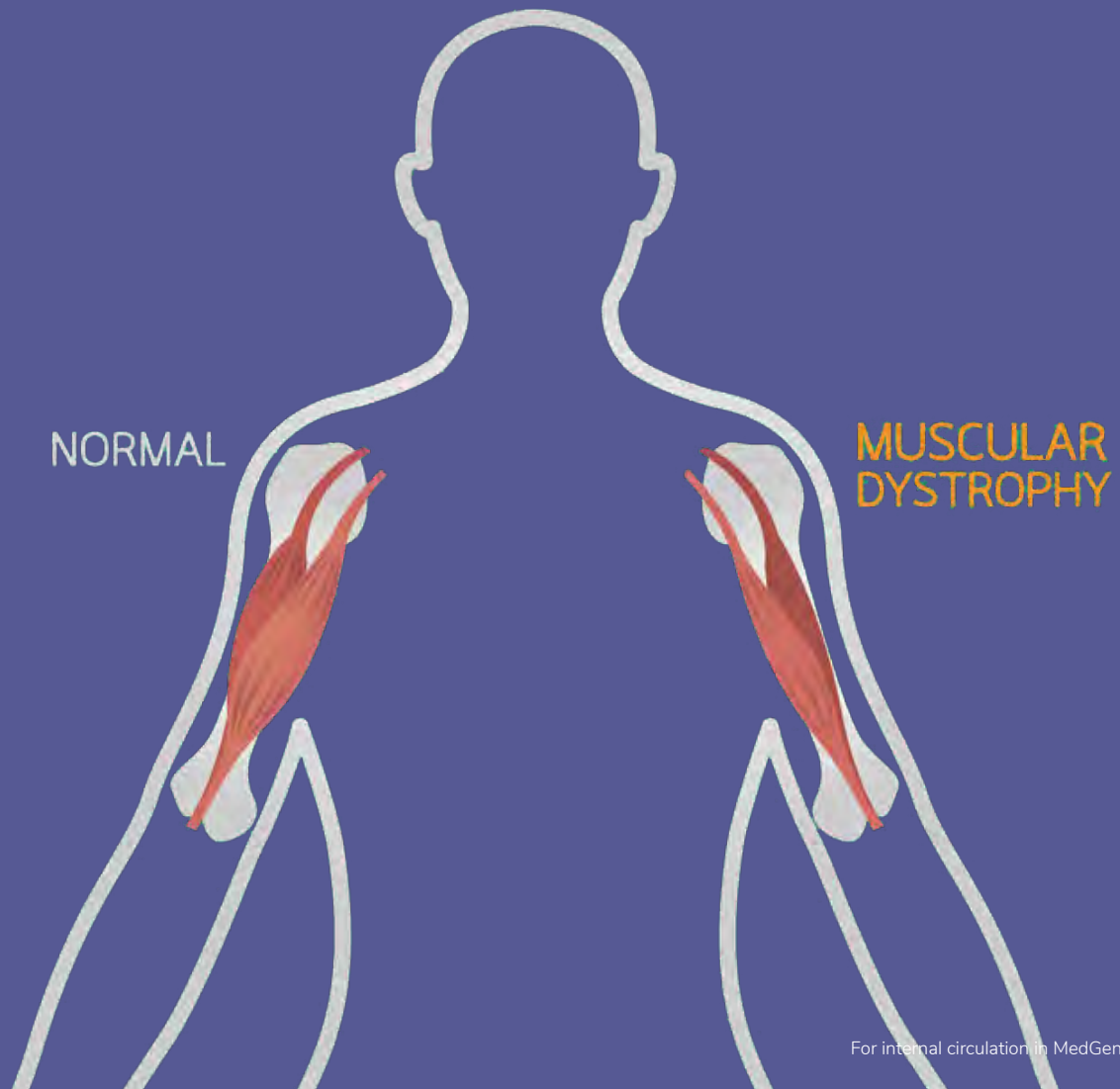
Duchenne Muscular Dystrophy (DMD) is one of the most common and devastating neuromuscular X-linked recessive genetic disorders affecting male children (Moser, 1984). DMD is caused by mutations in DMD (encoding dystrophin) that prevent the production of the muscle isoform of dystrophin (Dp427m). DMD spans a length of ˜2.5 Mb and comprises 79 exons and 78 introns. The DMD gene generates multiple transcripts encoding various dystrophin isoforms (e.g. lymphocyte dystrophin, cortical dystrophin, Purkinje dystrophin, foetal dystrophin, retinal dystrophin, muscle dystrophin, etc.). The muscle-specific isoforms transcribes a ˜14-kb long processed RNA that encodes a ˜427-kDa protein called 'Dystrophin' (G.recki et al., 1992; Koenig et al., 1988; Monaco et al., 1986). It is primarily expressed in skeletal, cardiac, and smooth muscles where it functions as a structural unit bridging both the internal actin cytoskeleton and the external sarcolemma forming a dystrophin–glycoprotein complex (Gao et al., 2016) that contributes to the structural integrity of the muscle fibre cells. The prevalence of DMD in male is less than 10 cases per 100,000. The distribution is identical between different parts of the world (Ryder, et al., 2017). In females DMD is very rare (<1 per million) and has been reported as individuals with Turner syndrome that involve translocation of DMD or with bi-allelic DMD mutations (Satre et al., 2004; Takeshita et al., 2017).

A doctor begins diagnosis of DMD by physical examination of the patient and understanding family history. Physical features like pseudohypertrophy, lumbar spine deviation, gait abnormalities, and several grades of diminished muscle reflexes indicates DMD. Further a doctor may observe change in an electrocardiogram. Apart from physical examination, doctors often recommend CK (creatine kinase) level blood test for diagnosis of DMD. Elevated CK level indicates that the muscle is being disintegrated by some abnormal process or inflammation. A very high level of CK indicates that the muscles are likely the cause of weakness and not the nerves that control them. High level of CK is found before the onset of DMD symptoms and the CK level peaks by the age of 2 years and then progressive falls when muscle tissues are replaced by fat and fibrotic tissues. Although high level CK indicates towards some form of muscular dystrophy but does not really confirm DMD. Some doctors can also recommend muscle biopsy to obtain more information. By examining the biopsy, doctors can tell what is happening to the muscles. The amount of functional dystrophin protein found in a muscle biopsy sample tell us more about the disease. When the sample fully lacks dystrophin protein it indicates DMD disease whereas when some functional dystrophin is identified then it indicates Becker muscular dystrophy (BMD). In modern era the biopsy is not recommended as nearly the patients undergo genetic testing for diagnosis of DMD.

Generally, patients with elevated serum CK levels are recommended to undergo genetic testing. Damaging mutation in the DMD gene confirms the disease. The diagnosis rate for DMD using genetic test is usually very high but, in some cases, where the genetic test turns out to be negative then western blotting or staining with selective antibody is done in the muscle biopsy. If dystrophin protein is detected in less than 5% of the normal quantity, then it indicates DMD.

The clinical course of DMD is remarkedly comparable from one patient to another. Most of the affected boys have retarded motor development and have difficulty in climbing stairs, and rise from the floor in a typical manner, by "climbing" up their legs. The key milestones of the diseases are loss of ambulation, scoliosis, ventilation, cardiomyopathy, and mortality. In 30% of the patient's loss of ambulation occurs by age of 10 years and by 15 years this increases to 90%. The mean age for the onset of scoliosis (a sideways curvature of the spine) is 14 years. The support for ventilation starts at the age of 15 and by 20 years of age up to half of the patients need ventilatory support. In 70% of the patient's cardiomyopathy occurs by the age of 15 years and in almost all patients by the age of 20 years. Up to 16% of patient die by age of 20 years and those surviving adulthood 60% succumbs to the disease by the age of 30% years (Szabo, 2021).



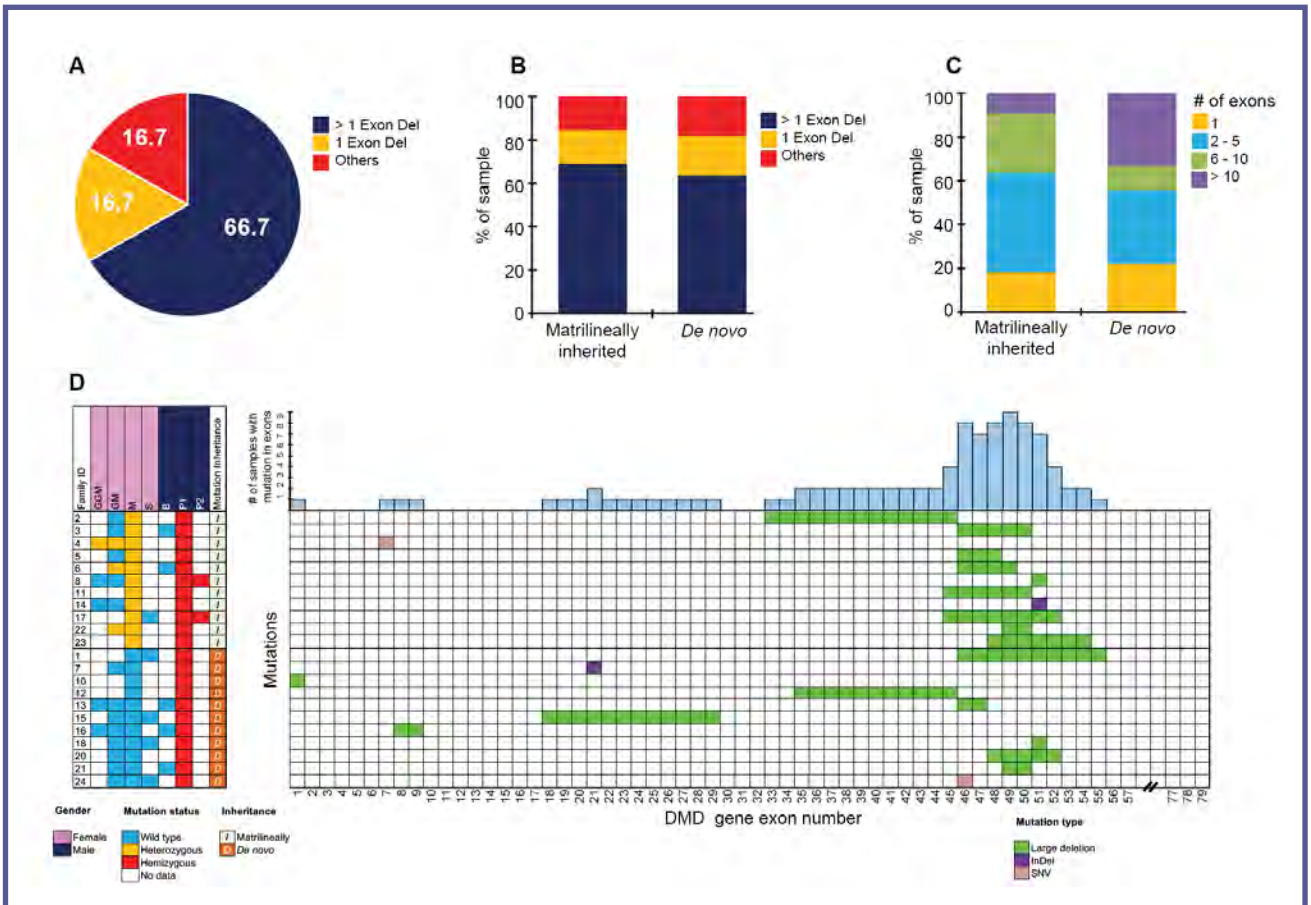


In about 60–65% of the DMD cases large deletions of one or more DMD gene exons (Dunnen, 1989; Elhawary *et al.*, 2018; Koenig et al., 1988) is observed. 20% are caused by single-nucleotide variations (SNV) including frameshift, nonsense, missense and short indel mutations (Aartsma-Rus *et al.*, 2006; Grimm *et al.*, 2012) and ~11% by duplications (Aartsma-rus *et al.*, 2016; White *et al.*, 2006). In general, the in-frame mutations cause a less severe form of muscular dystrophy, known as Becker muscular dystrophy (BMD), whereas frame-shift mutations lead to a more severe DMD phenotype (Muntoni et al., 2003). Majority of the mutations are frame-shift alterations that result in a prematurely truncated non-functional and unstable form of 'Dystrophin' protein product encoded by the DMD gene (Monaco *et al.*, 1988). The mutation rate in DMD is estimated to be higher than any other X-linked disorder (Winter & Pembrey, 1982), probably due to the size of the DMD gene. The mutation in DMD gene can occur due to maternal inheritance, a de novo event, or due to germline mosaicism. Globally several independent studies have been conducted to estimate the exact incidence of inherited and de novo DMD mutations. Studies have reported 16%–35% DMD patients with de novo mutation in the DMD gene (Aartsma-Rus *et al.*, 2012; Barbujani *et al.*, 1990; Caskey *et al.*, 1980; Garcia et al., 2014; Haldane, 1935). So far, no major genetic studies to understand inheritance patterns of DMD in South Asian context have been conducted. Moreover, to understand the inheritance of DMD gene variations, it is necessary to identify the exact genomic breakpoint to ascertain if the mutation is inherited or a *de novo* event involving the same exons.

As most common causal mutations for DMD are large deletions/duplications (copy number variation-(CNV)), multiplex ligation-dependent probe amplification (MLPA) is the current preferred diagnostic method (Abbs *et al.*, 2010; Bushby *et al.*, 2010; Falzarano *et al.*, 2015). MLPA is a multiplex PCR-based method that can detect deletions and duplications in patients or carriers (Lalic *et al.*, 2005; Verma *et al.*, 2012). Even though MLPA is a cost-effective method for diagnosis of CNV alterations in the DMD gene, it cannot give the genomic breakpoints of the deletion or duplication event. Also, MLPA has a higher propensity to miss small indels (<20 bp) and cannot detect single-nucleotide variants (SNVs) and deep intronic mutations (Aartsma-rus *et al.*, 2016; Prodduturi *et al.*, 2018). Patients showing symptoms but negative for DMD mutation using MLPA are tested using NGS techniques to detect SNVs or other small indels. Although the current NGS-based methods can detect exonic mutations and deletions, these cannot detect deep intronic variants or the associated genomic breakpoints deep inside the intronic regions. A single comprehensive genetic test that can detect all mutation types in the DMD gene would be an effective molecular diagnostic tool. It will provide a more accurate assessment of DMD carrier mutation prevalence in population.

At MedGenome, we designed a 2.08Mb (17,409 lockdown probes) DMD panel using set of oligonucleotide capture probes that spans the entire DMD gene including its 5′ UTR, 3′ UTR, 79 exons and 78 introns. The panel also covers 80 additional genes associated with different muscular dystrophies. Recently we published a study using this panel where we studied the inheritance of DMD genes across generations (Shastry *et al.*, 2020). We collected the matrilineal samples across four generations of 24 clinically confirmed DMD patients and from 22 unrelated families. Overall, our cohort had 77 subjects including the patients/probands and siblings from the current generation, and members of the maternal lineage (mother, grandmother, and great grandmother). All probands in the study were diagnosed with DMD based on established diagnosis criteria that included difficulty in walking, walking on toes, scoliosis, and frequent falls (Archer *et al.*, 2016). Patients in our studies have previously been confirmed positive for a DMD mutation by MLPA/mPCR and/or TrueSight gene panel, Illumina.

We generated ~500 Mb of data for each sample with >85% of bases passing the quality threshold of Q30 Phred score. 99.9% of reads mapped to the genome with a median read duplication rate of 15%. Overall, 79.54% of the DMD gene was captured by our panel, of which >94% was sequenced at an average depth of >200X. Using our clinically validated pipeline we identified SNVs and short InDels using Sentieon-GATK pipeline. We also used SoftSV and ExomeDepth programs to identify large deletions and duplications as it is critical for detection of larger deletion and duplication present in the DMD gene. Analysis of the DMD gene for pathogenic genetic alterations (SNPs, indels, large deletions) showed both maternally inherited (13 probands, 54%) and *de novo* (11 probands, 46%) mutations. As expected, the majority of the proband samples have loss-of-function mutations detected in DMD gene was due to large/single-exon deletions (83.3%; Fig 1a). Single-exon deletions were detected in 16.7% whereas multi-exon deletions in 66.7% of the proband samples. We found point mutation and a single-base indel in four proband samples (16.7%; Fig 1a). Eleven out of the 22 analyzed families showed maternally inherited DMD gene mutations. These mutations were hemizygous in the proband and heterozygous in the mother and other maternal subjects from the family. Nine of these inherited mutations were large deletions whereas two families showed single-nucleotide variations (SNV; Fig 1b). Of the total 11 probands with large, maternally inherited DMD deletions, we found 18.2%, 45.4%, 27.3%, and 9.1% to have 1 exon, 2–5, exons, 6–10 exons, and >10 exons long deletions, respectively (Fig 1c, d). Single-exon deletions were detected in two siblings who displayed a maternally inherited deletion of exon 51. Multiple-exon deletions were found to be inherited in eight families (Fig 1d). These deletions were found between exon 33. The largest deletion spanning 13 exons from exon 33 to 45. Deletions in four out of nine families were found only in the mother's sample whereas the grandmother or great grandmother did not show any detectable variants.



Overview of the mutations in the cohort: (a) Percentage of SNVs, single-exon deletions and large deletions identified in the cohort. (b) Percentage of sporadic (de novo) and maternally inherited mutations identified in the cohort. (c) Differential number of exons mutated in the de novo and inherited cases within the cohort. (d) Heatmaps summarizing the different DMD mutations (by type, inheritance, length and frequency) across the cohort.

De novo DMD mutations were identified in 11 probands of the 22 families. Of these nine families had large or single-exon deletions whereas those in two other families had SNVs. All the other maternal subjects in these families did not have any DMD mutations. The largest *de novo* detected from exon 18 to 29 and has not been reported previously. The distribution of the *de novo* large deletions across the exons was random and did not follow any specific pattern that could be explained as *de novo* mutation hotspot. Only one *de novo* SNV and one single-base deletion was identified in two unrelated families.

In our study sixty-five percent of the DMD mutations were found to be in the distal hotspot region between exons 45 and 55, 17% in the proximal region between exons 1 and 20, and the remaining 17% in the mid-region between exons 18 and 45. In the inherited mutational group, 9% (1/11) were found to occur in the proximal region, 82% (9/11) in the distal region, and 9% (1/11) in the mid-region, whereas in the *de novo* cases, the distribution of mutations was 25% (3/12) in the proximal region, 50% (6/12) in the distal region, and 25% (3/12) in the mid-region. However, distribution of point mutations with respect to the region was random and was found to be scattered among the distal and the proximal hotspot regions. No mutation was detected beyond exon 55 in the DMD gene.

DMD is still an incurable disease. Glucocorticoid therapy has been shown to delay the loss of ambulation in patients with DMD by 1–2 years. They also reduce the chance of scoliosis. Prednisone with a daily dose of 0.75 mg/kg or deflazacort with a daily dose of 0.9 mg/kg are the currently recommended drugs (Harneet Arora, 2019; Angelini & Peterle, 2012). The use of steroids and prednisolone versus deflazacort still remains controversial. In one of the previous studies, it was shown that treatment with deflazacort would result in delayed deterioration of ambulation and maintenance of key motor functions, when compared with prednisone (Shieh et al., 2018). But a study reported in India showed that prednisolone has more effects than deflazacort (Petnikota *et al*., 2016).

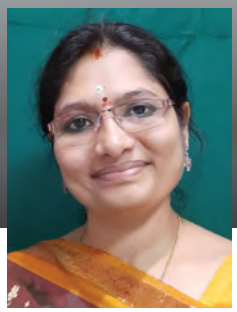
Several novel therapies targeting specific DMD mutations (including stop-codon read-through agents, exon-skipping antisense oligonucleotides (AONs) etc.) that target and restore Dystrophin function (Aartsma-Rus *et al.*, 2009; Babbs *et al.*, 2020; Haas *et al.*, 2015; Laing *et al.*, 2011; Ousterout *et al.*, 2015; Reinig *et al.*, 2017) have been developed. These drugs form a partially functional dystrophin and converts DMD to BMD phenotype. Successful results have been observed in mdx mice. Recently, FDA approved Amondys 45 (casimersen) injection developed by Sarepta Therapeutics for the treatment of Duchenne muscular dystrophy (DMD) in patients who have a confirmed mutation of the DMD gene in exon 45 and dystrophin function can partially be restored by skipping exon 45. Approximately 8% of patients with DMD can be treated using this drug. It has been estimated that approximately 70% of patients with deletions can be treated by single-exon skipping (Aartsma-Rus & Corey, 2020). Furthermore, it has been estimated that a larger number of patients carrying DMD duplications and nonsense mutations can be treated if multiple exons skipping is achieved (Aslesh et al., 2018; Dzierlega & Yokota, 2020).

DMD is not an easy disease to live with. In traditional Indian setting it is even more difficult to manage given that there are social taboos and absence of good infrastructure to help such patients. Getting a counselling support because of psychosocial issues makes it even worse for the families. There is an urgent need for increasing prenatal testing and carrier screening for high-risk mothers in India. Genetic counselling will play an important role in reducing the socioeconomic burden of the DMD disorder. There are also not many clinical trials going on in India. The pharmaceutical companies should be encouraged to conduct clinical trials in India. There has been considerable progress made in stem cell research in India, but more needs be done to fight this terrible incurable disease.

Featured article

Genetic Counseling in the times of Covid

From 'Why are you making lunch and breakfast together now' to 'Do you have to go to the hospital today?'



Madhavilatha, PhD,
Genetic Counsellor Level II

Work from Home-Genetic Counselling!!

Me: After a session on recurrent pregnancy loss

Daughter (curious): Maa, what is IVF?

Me: After a session on cancer

Daughter (hesitant): Maa, what is ovarian?

Me: After a session talking to parents

Daughter (anxious): Maa....what happened to the baby?

The first nationwide lockdown was announced by our honourable Prime Minister from March 25, 2020 onwards. There was fear, anxiety and worry for our near and dear ones. In May 2020, two months after the first lockdown was announced, I remember reading Dr. Ram's mail where among many other things he talked about the "new normal". COVID-19 or the SARS-CoV-2 virus created an upheaval in the lives of people, on both personal and professional levels. It challenged us in many ways to reconsider a wide variety of practices, one of which is driving people worldwide to work from home (WFH).

Work from Home

It is interesting to note that WFH is not new, and the concept was initially mentioned in 1970s as "telecommuting" or "telework". It has been used in various terms over the decades, namely remote work, flexible workplace, telework, telecommuting, e-working.

Many articles have been written talking about the advantages and limitation of WFH. The advantages, included are reduced commuting time, avoiding office politics, using less office space, improved gender diversity (eg. women and their careers), healthier workforces with less absenteeism, job satisfaction, better productivity and reduced work-life conflict.

Conversely, the drawbacks include lack of a good working environment, blurred line between work and family, inability of remote workers to disengage from work leading to overwork, social isolation, distracted by the presence of young children or family members.

The pandemic also saw a huge increase in the usage of telehealth services. A July 2021 article reported that the use of telehealth increased 38X from the pre-COVID-19 baseline.

Telemedicine refers specifically to remote clinical services, and the technology first began as a form of healthcare delivery in the late 1960s due to the needs of NASA! It is also effectively utilized in clinical genetics services - an application that has been termed "telegenetics." Telegenetics has been in practice for hereditary cancer, prenatal counseling, pediatric services and for a range of genetic disorders too, the benefits being convenience, reduced travel time and associated costs, and reduced waiting time to see a genetics specialist.

Studies in the pre-Covid era have reported high levels of patient satisfaction with the telegenetics services, as ascertained through questionnaires or interviews. Patients were generally satisfied with the technology used, the education and information provided, and the opportunity to communicate with genetics professionals without having to travel long distances. Patients tended to be more satisfied than the practitioners, viewed the technical difficulties involved as being less problematic, and were more satisfied with the rapport established with the genetics professional. There was a decrease in anxiety and depression after genetic counseling regardless of the counseling delivery method. However, the main difficulty was for the practitioner to detect nonverbal cues from patients.

Interestingly, MedGenome has been providing telegenetic services successfully for the past 5 years. Our main aim was to extend genetic counselling access to remote populations thereby increase the capacity to provide genetics services to those who require them and not just to those fortunate enough to live close to genetics centres. Of course, when we started, there were apprehensions from outside groups as to how genetic counselling can be provided online through telephone or video. But we have shown the way.

During the pandemic and the shift to tele-mode, for clinics that have only seen patients in-person, the transition to using telehealth presented a challenge. But due to our well-established telecounseling practices already in place, we were able to continue with our services and even provided, valuable guidance and support to other genetic counselors on how technology can be used to communicate with patients, deliver results, and provide support to patients.

Genetic counselling is well-suited to remote delivery via telephone or videoconferencing, as the clinical interaction consists largely of communication rather than physical examination. It can also allow use of the supportive tools such as information slides and brochures. Extensive use of videoconferencing during the pandemic has ushered in a new acceptance of virtual meetings. While a person on the telephone enjoys the convenience of the experience, a person/family in the video would be able to see the counselor's body language as well.

Personal Experience as a genetic counsellor - WFH and Hospital visits

Work from home requires a quiet and dedicated space to perform work duties, which can be a real challenge especially for those whose work involves talking and listening to patients. We cannot work listening to music or a TV in the background. Even small sounds are a distraction.

During lockdown, family members could not understand as to why breakfast and lunch was being prepared together as usual. It was difficult to explain that we had to login on time and that it is not vacation time.

Very quickly our logistics was able to arrange for sample pickup three days a week, and I was going to the hospital to provide counselling especially for prenatal, neonatal and oncology cases. The floor in which the genetic counselling room was situated, was taken up for setting up the RT-PCR facility. So, counselling had to be done in any available room that day.

Come 2021, the hospital visits became more regular and more extended. And of course, we too had our share of Covid positive status in the family, twice.

Covid Gifts- a cloud with silver lining?

Amidst the uncertainties and chaos created by covid, one can still look into a few positive experiences of the lockdown such as the clean and clear roads. The 13km drive which used to take an hour earlier with the traffic, was now the most blessed for me. As the only person stepping out, I took on the responsibility of bringing groceries for my family as well as for a few neighbours.

Understanding the Covid situation might not get better enough for children to go to school for another year, we felt that it was the right time to get a pet that would incorporate the feeling of nurture and compassion in kids.

Covid-an eye opener

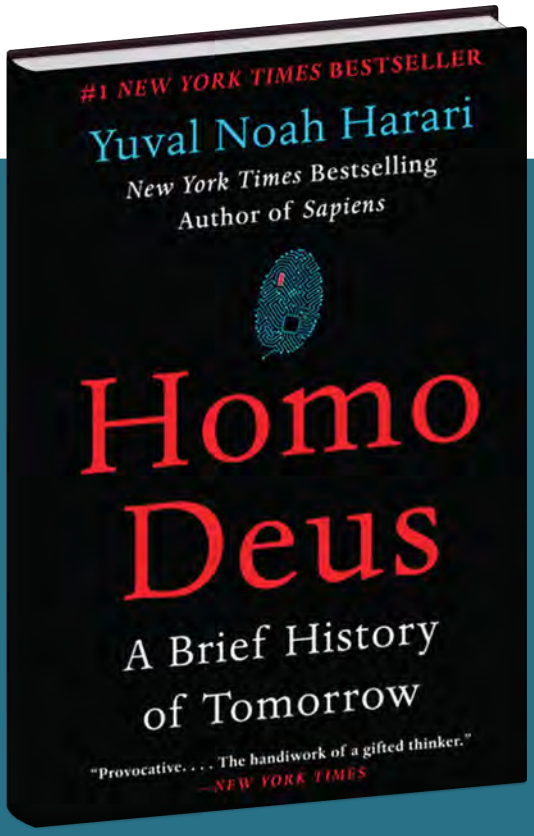
Gender equality always have been a bone of contention in every society. Even in covid times, reports state that woman and mothers have felt the most negative impact of remote working. This is because even when both partners are working from home, women have to handle a majority share of domestic responsibilities. Though there have been numerous WhatsApp videos/memes showing men cutting vegetables, there is still a great need to sensitise men they need to share the household chores, and it should be done every day!

The second and most disturbing is the psychology of the children. They got used to parents especially their mothers being at home. With parents leaving for work while schools are yet to start, has taken a huge toll on kids. For the past few months every morning the first thing my daughter invariably asks me is, 'Are you going today' or 'Do you have to go today?' The major issues seen are increased screen time, not attending classes in the absence of parents, untimely food, loss of handwriting skills, absence of face-to-face socialising, less behavioural development and many more, only time will tell.

The pandemic has touched every one of us in different ways in both our personal and professional lives. Its impact has been both positive and negative. As genetic counsellors we strive to make a difference in someone's life in our own way and tele-genetics is here to stay.

Lokah Samastah Sukhino Bhavantu

From our Colleagues



Book Review

Book

Homo Deus -

A brief history of tomorrow

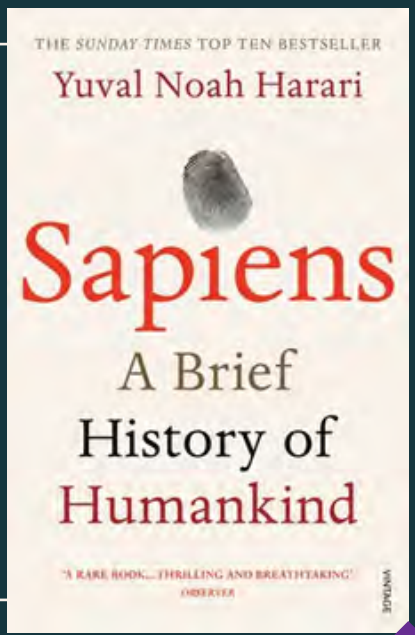
Book review by

Dr. Ramesh Menon,

Associate Director, MedGenome Labs, Bengaluru

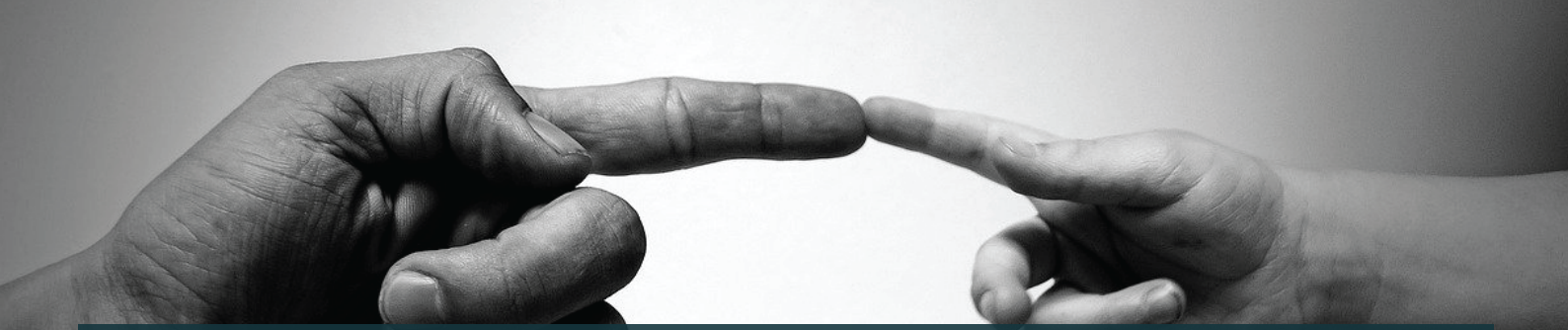
From my early school days, I was keen in reading; from Malayalam kids' comic magazine such as Balarama, like every 80's kid who grew up in Kerala. In my later school days, I was interested in reading stories behind scientific discoveries, stories about kings, autobiographies, and evolution (in a broader sense). I was an active book reader till about 2010, but now my free time goes into news feeds, reading scientific articles. And unfortunately, the time for book reading has significantly reduced, thanks to the mobile phone! The last book I read (completed) was a year ago. So, excuse me for reviewing a 5-year-old book.

Homo Deus is an interesting book by Yuval Noah Harari (Publisher: Harvill Secker, 2016), which talks about similar subject that of his previous book, Sapiens. In fact, it is an extension of Sapiens, which describes about human history that brought up us to the present day. If you've read Sapiens, the first two parts of Homo Deus will sound quite similar (and less engaging).



The first part of the book talks about how humans overcome famine, infectious diseases, and violence.

Though these exists in several parts of the world even now, but the damage caused by these are way smaller than lifestyle diseases such as obesity and diabetes for instance. As a result of scientific advancements, human lifespans could double and more. Humans have conquered the world, as rise from an insignificant animal of 200,000 years ago to our current dominant position. For instance, proportion of domesticated animals (farmed or companion animals) compared to wild animals shows how human took control of the living world in these years, and the proportion of wild animals/birds are still declining. When humans were hunter/gatherers, they lived as a part of nature, but the evolving intelligence separated human from other animals and over-powered, controlled and used them for self-growth.



This not only restricts using them as a source of food, but also animals are used for pharmacological testing in labs for the development of new drugs for our well-being. One of the unique ability of humans is the capability to interact with very large number of individuals (enormous socialization), which is key to our mastery on the planet. The second part of the book describes how we give meaning to the world. Humanism is a belief that is the ultimate determination of any act to be right and wrong lies in whether that act makes people feel better or happy. The author says the future religion will be Humanism.

The third part is the real core of the book, describes how homo sapiens will be replaced by homo deus (Sapiens=wise, Deus=god). We will constitute a new species with the help of genetic engineering, artificial intelligence, which will render homo sapiens as an irrelevant resource, except as we have value to the new master species. All our actions and thought processes are results of data-driven complex algorithms work within ourselves, contributed by genetics and life experiences. We ignore the pain caused by animal, if we think the suffering is not related to a higher intelligence. This will become true for us too. The new world of Homo Deus getting created, which will supress and replace Homo sapiens.

Overall, Homo Deus describes how humans conquered and controlled the world, where we are now, through agricultural and industrial revolutions. The new revolution is based on data, algorithms and devices, through which we dictate and facilitate our own divine nature.

The book is engaging with some interesting fact-based logical assumptions and brings light to the certain faculties of mind such as, imagination and perception about development of human race. Homo Deus has lot of overlap with the Sapiens, which is quite repetitive. It has slight moral or philosophical angles too, which I didn't enjoy much. Nevertheless, for those who are interested to know how we have evolved, and where we are moving to, go for it!

From our Colleagues

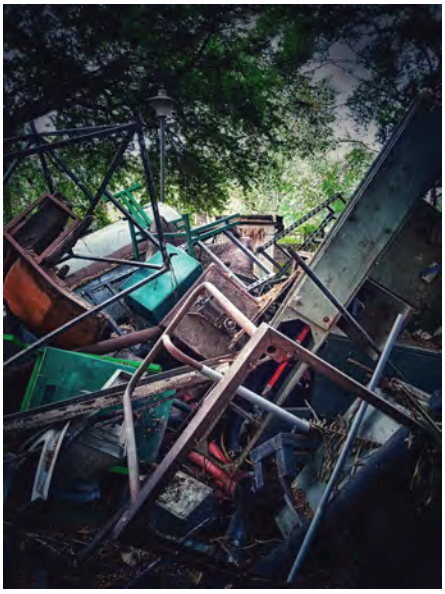
Art meets Science

Man is unique not because he does science, and he is unique not because he does art, but because science and art equally are expressions of his marvelous plasticity of mind. — Jacob Bronowski

By: Raseed I Munawalli,
Lead Research Associate

By: Smitha Sandheep,
Senior Research Associate

From our Colleagues



By: Sharath Basavaraju,
Research Associate Trainee

From our Colleagues

Our employee's little Picasso :)





By
K. Warrier (12 years)
DNA of Hiranjith GH

By
Varunkrishna, (10yrs)
DNA of Dr E Venkataswamy

By
Gurucharan, (7 yrs)
DNA of Dr E Venkataswamy

Employee Connect

1. Gregor Mendel's discovery of basic hereditary principles was not realized until the early 20th century. Who were the scientists who rediscovered his work?

   a) Francis Crick, James Watson, and Rosalind Franklin b) Hugo de Vries, Carl Correns, and Erich Tschermak von Seysenegg

   c) Marie Curie and Irène Curie d) Charles Darwin and Herbert Spencer

2. Who provided the first conclusive evidence, in 1902, that chromosomes carry the units of inheritance and occur in distinct pairs?

   a) Walter Sutton b) Barbara McClintock c) Clarence E. McClung d) Christiane Nüsslein-Volhard

3. In 1944 which American biologist, along with Oswald T. Avery and Colin M. MacLeod, used bacteria to provide the first experimental evidence that the genetic material of living cells is composed of DNA?

   a) Ernst Mayr b) Frederick Sanger c) Maclyn McCarty d) Paul Alfred Weiss

4. Who deduced that the sex of an individual is determined by a particular chromosome?

   a) Nettie Maria Stevens b) Theodor Schwann c) A.D. Hershey d) Sylvia Earle

5. Who laid the mathematical foundation of the science of genetics?

   a) Gregor Mendel b) Carl Correns c) Dorothy Hodgkin d) Erich Tschermak von Seysenegg

6. Who laid the foundation for James Watson and Francis Crick to suggest the helical structure of DNA?

   a) Lise Meitner b) Alan Hodgkin c) Rosalind Franklin d) Peter B. Medawar

7. Which of these congenital disorders is characterized by an extra chromosome?

   a) Down syndrome b) trisomy 13 c) Turner syndrome d) heart disease

Previous puzzleWinner

Anshu,
Genome Analyst

*Kindly mail your answers by 15th November 2021 to **editor@medgenome.com**. The first two people to answer the quiz correct will be featured in the next edition of our newsletter.*

Answer the Quiz & WIN Voucher

Employee Connect

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Kishore


Sinod


Ganapathi Bhat


Devendra Kumar Sumeriya


Swaraj Paul


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Sandeep Bhakare


Vivek Singh


Jitendra Keshri


Sneha Jaiswal


D M Pavan


Priya Bharti


Kiran Sony


Mohammed Shabbir Rampurawala


Avula Praveen Reddy


Saleemali Kutubuddeen Nadaf


Naushad Ansari


Puneeth H S


Ajay Mohan Gupta


Deeksha Chugh


Benny T K


Nishag


Indrabhushan


Nitin Vyas


Amit Gupta


Tehmin Raj


Baby Boruah


Jerina Doley


Ashwini N


Nalesh


Akarsh


Pavan


Murali


Balasatheesh


Tushar


Abimanyu


Abhishek Kumar


Soumen


Abhishek mayekar

Senthil Kumaran

Rahul Bhagat

Sumit

Helen

Divya

Mashoor

Partheeban

Jayanthan

Shivakumar

Photo Feature

Celebrations

'Freedom of expression' through talent display by our colleagues. Just look at the creative talents we have with us!!


Of course, there was a tricolor fiesta in our offices!!





The glorious Onam festival was celebrated with the customary rangoli making and ethnic wear.




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Wishes You and Your Family a Happy Diwali!

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